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ABSTRACT

Fragile X syndrome is an inherited genetic condition identified by a break or a weakness on the long arm of the X chromosome, and associated with mental retardation. Described on this single-sheet information guide are the prevalence of fragile X syndrome, its increased rate of incidence in boys as compared to girls, physical characteristics, degree of associated mental retardation, behavioral symptoms, treatment, historical identification, chances of hereditary transmission, resources for genetic counseling, and a list of references. (JDD)

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Association for Retarded Citizens

FRAGILE X SYNDROME

What is Fragile X Syndrome?

Fragile X syndrome is an inherited genetic condition associated with mental retardation. It is identified by a break or a weakness on the long arm of the X chromosome. Since this is an abnormality of a sex chromosome which can be transmitted from parent to child, the term "sex-linked" or "X-linked" inheritance is used in the medical literature. This means that mothers are carriers and their sons are at risk of being affected, while their daughters are at risk of being carriers and sometimes mildly affected. While more boys than girls are affected by fragile X, it is not transmitted from father to son.

How Common is Fragile X Syndrome?

Since no large-scale surveys of newborns have been carried out, estimates vary on the prevalence of fragile X syndrome. An evaluation of school children in England found a prevalence of one out of 1,350 males and one out of 2,033 females with fragile X mental retardation, giving a total prevalence of the fragile X "gene" of one per 1,634 in the school children studied (Webb, 1986). If a three percent prevalence of mental retardation in the general population is used, fragile X syndrome may account for up to ten percent of all mental retardation. It is second to Down syndrome among genetic causes of mental retardation (Rogers and Simensen, 1987).

Why Aren't Girls Affected as Much as Boys?

The X chromosome is unique among the 23 pair of chromosomes found in each human cell. It is one-half of the chromosome pair that determines sex. If, at conception, an embryo receives an X chromosome from the mother and a second one from the father, it will be female. But if it gets a Y-shaped chromosome from the father, the embryo will be male. Thus, males can only inherit fragile X from their mothers.

Disorders related to an imperfect X chromosome usually don't show up in females because they have a second, presumably normal one. Males usually do suffer effects, as they have just the one X chromosome (Bishop, 1986).

What Are the Physical Characteristics of Fragile X Syndrome?

Most children with fragile X appear physically normal at first. As they grow, males may show facial features that can be recognized about half the time. These features include a long narrow face and prominent ears, jaw and forehead. Enlarged testicles (known as macro-orchidism) is another physical characteristic.

Physically distinct features which can aid in diagnosis are sometimes present in affected females (Turner, *et al*, 1986). Various researchers have noted a long face, protruding jaw,

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prominent forehead, prominent or cupped ears and hyperextensible finger joints. However, because physical features are subtle and not present in many females, they cannot be relied on to document their carrier status (Hagerman, 1987).

What Degree of Mental Retardation is Associated with Fragile X Syndrome?

About 80 percent of boys who inherit fragile X have mental impairment, ranging from severe retardation to low-normal intelligence. The majority are mildly to moderately retarded. Girls are much less affected, with estimates that about 30 percent with the genetic condition have some degree of mental retardation (Sherman, *et al*, 1985).

What Behavioral Symptoms are Associated with Fragile X?

Men and boys with the fragile X syndrome are usually socially engaging, but they have an unusual style of interacting with other people. They tend to avoid direct eye contact during conversation, and handflapping or handbiting is common. They may have an unusual speech pattern characterized by a fast and fluctuating rate and repetitions of sounds, words or phrases. They also may have problems in their attention span, hyperactivity and motor delays. Some males demonstrate autistic-like behaviors, including perseverative speech, unusual hand mannerisms and problems in relating to others (Hagerman, 1987).

Can Fragile X Be Treated?

There is no cure for fragile X syndrome, but medical intervention can improve the problems in attention and hyperactivity of many young boys (Hagerman, 1987).

Treatment for children with fragile X syndrome is determined by an individualized program of special education including such components as speech therapy, physical therapy and vocational preparation. A child less than age three can benefit from an early intervention program. In general, children who are living at home and enrolled in special education will function at a higher level as adults than do men who were institutionalized years ago and never received special education.

When Was Fragile X Syndrome Identified?

Although it was well-known for years that more males than females are affected by mental retardation, it was the 1960s before scientists perfected ways to look at chromosomes and detect abnormalities.

The fragile site on the long arm of the X chromosome was first noted in 1969 by Dr. Herbert A. Lubs who discovered these chromosomes in a family with two brothers with mental retardation. By the late 1970s this condition was named fragile X by Dr. Grant R. Sutherland who was studying the occurrence of fragile sites. While the announcement about the fragile X chromosome was first published in the medical press in the late 1970s, it has only come to the attention of the public in the last five years. Because of its recent

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discovery, most individuals with this syndrome are undiagnosed.

What are the Chances of Having a Child with Fragile X?

To date, no one model for the transmission of fragile X has been proven and accepted. As a result, predictions of transmission from parent to child are not fully accurate. However, some general guidelines may be given. Either the father or the mother must have visible signs of the syndrome or must be a carrier of the syndrome in order to transmit fragile X. In the case of fragile X, carriers are usually females who possess the fragile X chromosome but do not show visible outward signs of the syndrome. An estimated one out of three female carriers is mentally affected.

- A male child has a one out of two (50 percent) chance of being affected by fragile X if his mother is either a carrier or has fragile X.
- A male child has no chance of being affected by fragile X if his father has fragile X and his mother does not.
- A female has a 50 percent chance of being a carrier if her mother is a carrier or has fragile X.
- A female will be a carrier if her father has fragile X.

Due to the presence of suspected silent male carriers, genetic analysis can become very complicated. Therefore, in order to obtain a more accurate prediction of the chances of having a child with fragile X syndrome, genetic counseling should be obtained.

Where Can Information About Genetic Counseling be Obtained?

The National Center for Education in Maternal and Child Health (NCEMCH) is a major link between sources of information, services and the professional in areas of maternal and child health, including medical genetics. It provides

numerous services including responding to requests for information, maintaining a resource center of educational materials, and publishing a variety of guides, directories, bibliographies and newsletters. It publishes *A Guide to Selected National Genetic Voluntary Organizations*. NCEMCH's address is 3520 Prospect St., N.W., Washington, D.C. 20057.

A national Fragile X Foundation also exists which provides educational materials and links families to parent support groups. Its address is P.O. Box 300233, Denver, Colorado 80203.

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